

AMENDMENTS TO THE CLAIMS

1. (Currently Amended) A method of diagnosing myelinopathy in an individual, said myelinopathy resulting from a periaxin alteration in the individual, comprising the steps of:

obtaining a sample containing nucleic acid from said individual;

assaying said sample for an alteration in a periaxin polynucleotide, ~~wherein said alteration is associated with said myelinopathy.~~

2. (Previously Presented) The method of claim 1, wherein said periaxin polynucleotide is SEQ ID NO:76.

3. (Original) The method of claim 1, wherein said periaxin polynucleotide is SEQ ID NO:1, SEQ ID NO:27, SEQ ID NO:28, SEQ ID NO:29, SEQ ID NO:30, SEQ ID NO:31, SEQ ID NO:32, SEQ ID NO:33, SEQ ID NO:34, SEQ ID NO:35, SEQ ID NO:36, SEQ ID NO:37, SEQ ID NO:38, SEQ ID NO:39, SEQ ID NO:40, SEQ ID NO:41, SEQ ID NO:42, SEQ ID NO:43, SEQ ID NO:44, SEQ ID NO:45, SEQ ID NO:46, SEQ ID NO:47, SEQ ID NO:48, SEQ ID NO:49, SEQ ID NO:50, SEQ ID NO:51, SEQ ID NO:52, SEQ ID NO:53, SEQ ID NO:54, SEQ ID NO:55, SEQ ID NO:56, SEQ ID NO:57, SEQ ID NO:58, SEQ ID NO:59, SEQ ID NO:60, SEQ ID NO:61, SEQ ID NO:62, SEQ ID NO:63, SEQ ID NO:64, SEQ ID NO:65, SEQ ID NO:66, SEQ ID NO:67, SEQ ID NO:68, SEQ ID NO:69, SEQ ID NO:70, SEQ ID NO:71, SEQ ID NO:72, SEQ ID NO:73, SEQ ID NO:74, SEQ ID NO:75, SEQ ID NO:76, or SEQ ID NO:77.

4. (Original) The method of claim 1, wherein said myelinopathy is selected from the group consisting of Charcot-Marie-Tooth (CMT) syndrome, hereditary neuropathy with liability to pressure palsies (HNPP), Dejerine-Sottas syndrome (DSS), congenital hypomyelinating neuropathy (CHN), and Roussy-Levy syndrome (RLS).

5. (Original) The method of claim 1, wherein said assaying step further comprises a polymerase chain reaction.

6. (Original) The method of claim 5, wherein primers for said polymerase chain reaction are selected from the group consisting of SEQ ID NO:3, SEQ ID NO:4, SEQ ID NO:5, SEQ ID NO:6, SEQ ID NO:7, SEQ ID NO:8, SEQ ID NO:9, SEQ ID NO:10, SEQ ID NO:11, SEQ ID NO:12, SEQ ID NO:13, SEQ ID NO:14, SEQ ID NO:15, SEQ ID NO:16, SEQ ID NO:17, SEQ ID NO:18, SEQ ID NO:19, SEQ ID NO:20, SEQ ID NO:21, SEQ ID NO:22, SEQ ID NO:23, SEQ ID NO:24, SEQ ID NO:25, and SEQ ID NO:26.

7. (Previously Presented) The method of claim 1, wherein said alteration is 3775G>A, 1216G>A, 4075-4077d, 1483G>C, 3394A>G, 3248C>G, 2763A>G, 2645C>T, 306C>T, 1491C>G, 2655T>C, 2145T>A, 1102C>T, 2289delT, 2787delC, 2857C>T, or 247ΔC.

Claims 8-34. (Previously Cancelled)

35. (Previously Presented and Currently Amended) A method of detecting the presence or absence of a mutation associated with a myelinopathy, said myelinopathy resulting from a periaxin mutation in the individual, the method comprising:

- a) isolating a test nucleic acid from a subject, said test nucleic acid comprising a periaxin polynucleotide;
- b) comparing the test nucleic acid to a reference wild-type periaxin polynucleotide; and
- c) determining the differences between the test nucleic acid and the reference wild-type periaxin polynucleotide, wherein the differences are mutations in the periaxin polynucleotide of the subject, ~~and wherein the presence of a mutation in the periaxin polynucleotide of the subject is associated with the myelinopathy in the subject.~~

36. (Previously Presented) The method of claim 35, wherein said mutation is 2145T>A, 1102C>T, 2289delT, 2787delC, 2857C>T, or 247ΔC.

37. (Previously Presented) The method of claim 35, wherein said mutation encodes a defect of a periaxin polypeptide, wherein the defect is R953X, R368X, S929fsX957, R196X, V763fsX774, C715X, or R82fsX96.

38. (Original) The method of claim 35, wherein said periaxin polynucleotide is SEQ ID NO:1, SEQ ID NO:27, SEQ ID NO:28, SEQ ID NO:29, SEQ ID NO:30, SEQ ID NO:31, SEQ ID NO:32, SEQ ID NO:33, SEQ ID NO:34, SEQ ID NO:35, SEQ ID NO:36, SEQ ID NO:37, SEQ ID NO:38, SEQ ID NO:39, SEQ ID NO:40, SEQ ID NO:41, SEQ ID NO:42, SEQ ID NO:43, SEQ ID NO:44, SEQ ID NO:45, SEQ ID NO:46, SEQ ID NO:47, SEQ ID NO:48, SEQ ID NO:49, SEQ ID NO:50, SEQ ID NO:51, SEQ ID NO:52, SEQ ID NO:53, SEQ ID NO:54, SEQ ID NO:55, SEQ ID NO:56, SEQ ID NO:57, SEQ ID NO:58, SEQ ID NO:59, SEQ ID NO:60, SEQ ID NO:61, SEQ ID NO:62, SEQ ID NO:63, SEQ ID NO:64, SEQ ID NO:65, SEQ ID NO:66, SEQ ID NO:67, SEQ ID NO:68, SEQ ID NO:69, SEQ ID NO:70, SEQ ID NO:71, SEQ ID NO:72, SEQ ID NO:73, SEQ ID NO:74, SEQ ID NO:75, SEQ ID NO:76, or SEQ ID NO:77.

39. (Original) The method of claim 35, wherein said comparing step is by DHPLC, sequencing, hybridization, or a combination thereof.

40. (Original) The method of claim 35, wherein the myelinopathy is Charcot-Marie-Tooth (CMT) syndrome, hereditary neuropathy with liability to pressure palsies (HNPP), Dejerine-Sottas syndrome (DSS), congenital hypomyelinating neuropathy (CHN), or Roussy-Levy Syndrome (RLS).

41. (Previously Presented and Cancelled Herein).

42. (Previously Presented) The method of claim 35, wherein said mutation encodes a defect of a periaxin polypeptide, wherein the defect is E1259K, A406T, E1359del Δ , E495Q, R1132G, P1083R, I921M, A882V, T102T, P497P, or P885P.

43. (New) A method of diagnosing myelinopathy in an individual comprising the steps of:

obtaining a sample containing nucleic acid from said individual;

assaying said sample for an alteration in a periaxin polynucleotide, wherein said alteration is associated with said myelinopathy, and wherein said myelinopathy comprises a prominent sensory neuropathy.

44. (New) The method of claim 43, wherein said periaxin polynucleotide is SEQ ID NO:76.
45. (New) The method of claim 43, wherein said periaxin polynucleotide is SEQ ID NO:1, SEQ ID NO:27, SEQ ID NO:28, SEQ ID NO:29, SEQ ID NO:30, SEQ ID NO:31, SEQ ID NO:32, SEQ ID NO:33, SEQ ID NO:34, SEQ ID NO:35, SEQ ID NO:36, SEQ ID NO:37, SEQ ID NO:38, SEQ ID NO:39, SEQ ID NO:40, SEQ ID NO:41, SEQ ID NO:42, SEQ ID NO:43, SEQ ID NO:44, SEQ ID NO:45, SEQ ID NO:46, SEQ ID NO:47, SEQ ID NO:48, SEQ ID NO:49, SEQ ID NO:50, SEQ ID NO:51, SEQ ID NO:52, SEQ ID NO:53, SEQ ID NO:54, SEQ ID NO:55, SEQ ID NO:56, SEQ ID NO:57, SEQ ID NO:58, SEQ ID NO:59, SEQ ID NO:60, SEQ ID NO:61, SEQ ID NO:62, SEQ ID NO:63, SEQ ID NO:64, SEQ ID NO:65, SEQ ID NO:66, SEQ ID NO:67, SEQ ID NO:68, SEQ ID NO:69, SEQ ID NO:70, SEQ ID NO:71, SEQ ID NO:72, SEQ ID NO:73, SEQ ID NO:74, SEQ ID NO:75, SEQ ID NO:76, or SEQ ID NO:77.
46. (New) The method of claim 43, wherein said assaying step further comprises a polymerase chain reaction.
47. (New) The method of claim 46, wherein primers for said polymerase chain reaction are selected from the group consisting of SEQ ID NO:3, SEQ ID NO:4, SEQ ID NO:5, SEQ ID NO:6, SEQ ID NO:7, SEQ ID NO:8, SEQ ID NO:9, SEQ ID NO:10, SEQ ID NO:11, SEQ ID NO:12, SEQ ID NO:13, SEQ ID NO:14, SEQ ID NO:15, SEQ ID NO:16, SEQ ID NO:17, SEQ ID NO:18, SEQ ID NO:19, SEQ ID NO:20, SEQ ID NO:21, SEQ ID NO:22, SEQ ID NO:23, SEQ ID NO:24, SEQ ID NO:25, and SEQ ID NO:26.
42. (New) The method of claim 43, wherein said alteration is 3775G>A, 1216G>A, 4075-4077d, 1483G>C, 3394A>G, 3248C>G, 2763A>G, 2645C>T, 306C>T, 1491C>G, 2655T>C, 2145T>A, 1102C>T, 2289delT, 2787delC, 2857C>T, or 247ΔC.

49. (New) A method of detecting a polymorphism or mutation in a periaxin polynucleotide of an individual, comprising the steps of:
- obtaining a sample comprising said periaxin polynucleotide from said individual;
- assaying said periaxin polynucleotide for the polymorphism or mutation.
50. (New) The method of claim 49, wherein said periaxin polynucleotide comprises SEQ ID NO:76.

INTERVIEW SUMMARY

A telephonic interview in this application occurred on November 14, 2003 and included Applicants' representatives Thomas D. Paul and Melissa L. Sistrunk, and Examiner Chunduru. No exhibit was provided.

Claim 1 as rejected under 35 U.S.C. §112, first paragraph rejection was discussed, particularly regarding enablement of the claim in view of the teachings of the specification. In particular, Applicants' representatives argued that not all myelinopathies were encompassed in the claims but that only those myelinopathies having a periaxin mutation would be diagnosed. Examiner Chunduru expressed concern regarding enablement and inheritance patterns related to the myelinopathies, and Applicants' representatives argued that the inheritance pattern of the myelinopathies was irrelevant, so long as the myelinopathy resulted from a periaxin defect. Furthermore, they argued that a skilled artisan could identify inheritance patterns related to the mutation from general knowledge in the art. Agreement was not reached regarding the rejection, although amendments to overcome the rejection were discussed.